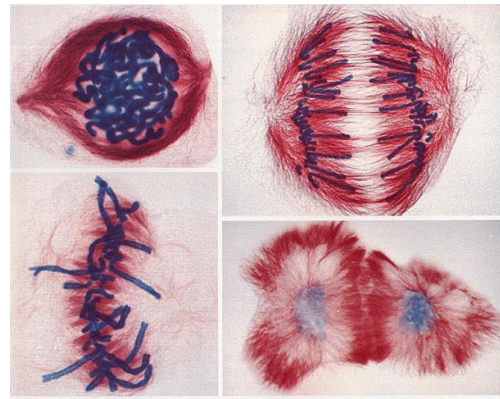


Genetic Disorders During Meiosis

Karyotypes

Genetic Technologies



Learning goals

- **Understand the errors that can occur during meiosis and identify some disorders using karyotypes**
- **Understand Mendel's 2 laws (Segregation and independent assortment)**
- **Understand the use of several genetic technologies**

Genetic Disorders

- **Clinical health problems visible at birth are called congenital defects**
- **They are caused by mutations in genes or environmental agents**
- **Some example of environmental agents that can affect a baby at birth are:**
 - Alcohol abuse by the mother during pregnancy
 - Contracting certain viruses (German measles) while pregnant

Genetic Disorders - Chromosomal

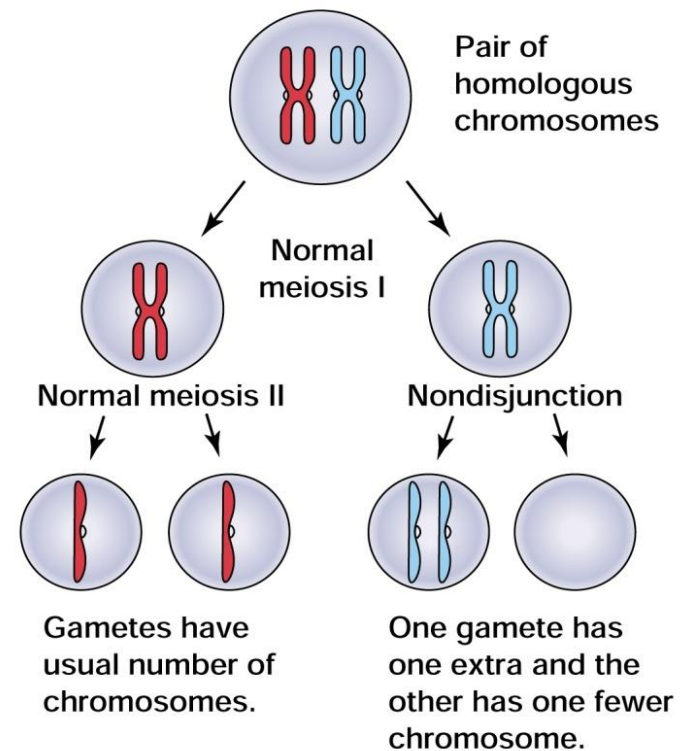
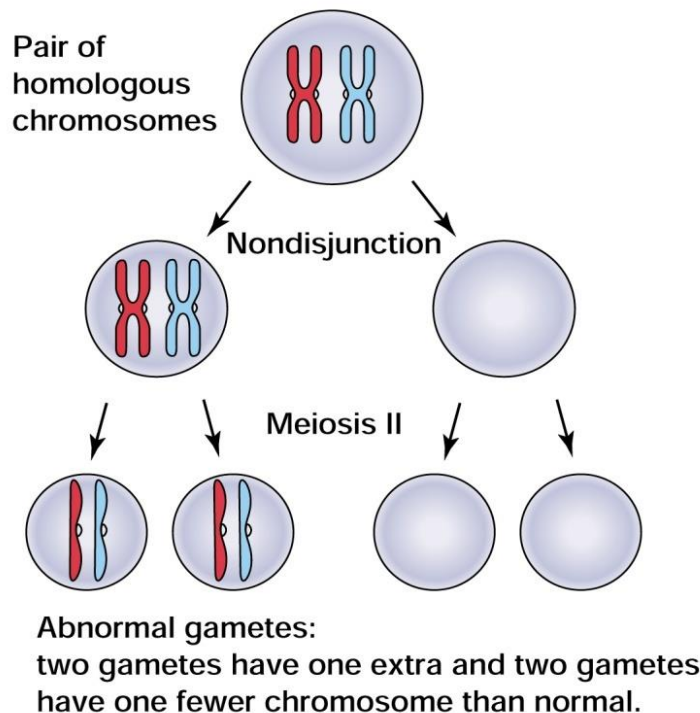
- Other genetic defects occur when chromosomes line up in meiosis in ways other than the usual pairings (e.g., the chromosomes don't split)
- This is called **meiotic non-disjunction**
- **Down Syndrome** is an example of a chromosomal genetic disorder. Individuals with down syndrome have an extra chromosome 21.

ERRORS IN MEIOSIS

- **Changes, or mutations in chromosomes can have severe consequences.**
- **Because chromosomes are copied during *interphase*, all daughter cells (sperm or egg) will carry the mutation.**
- **If that sperm or egg is part of fertilization, the new organism will carry that error in all of its cells.**

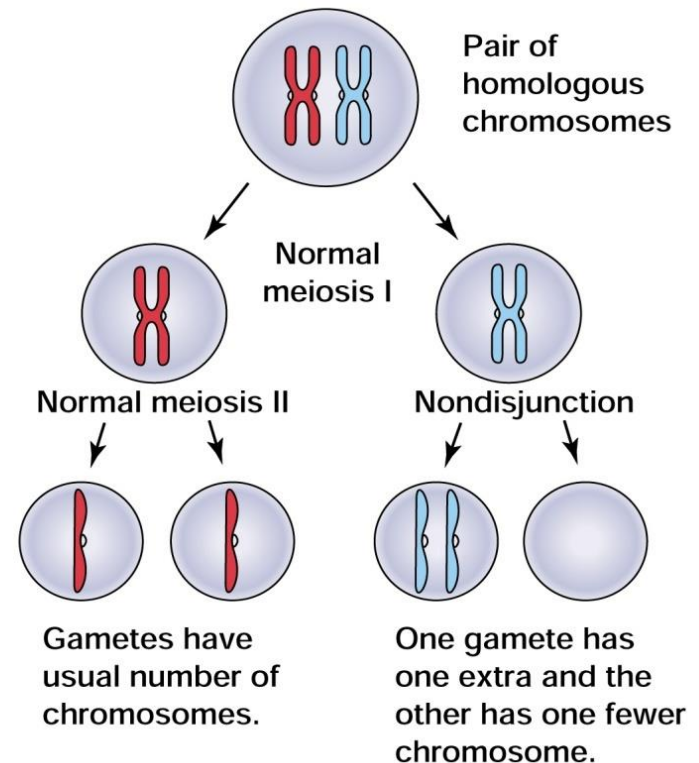
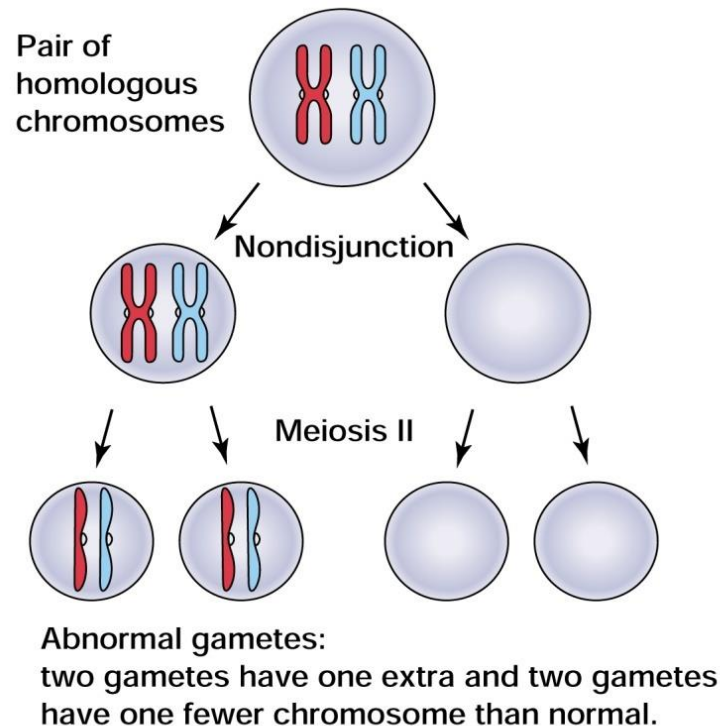
NON-DISJUNCTION

- The failure of chromosomes or tetrads to separate properly during anaphase is called **non-disjunction**. This results in the **addition** or **deletion** of a chromosome in a gamete.



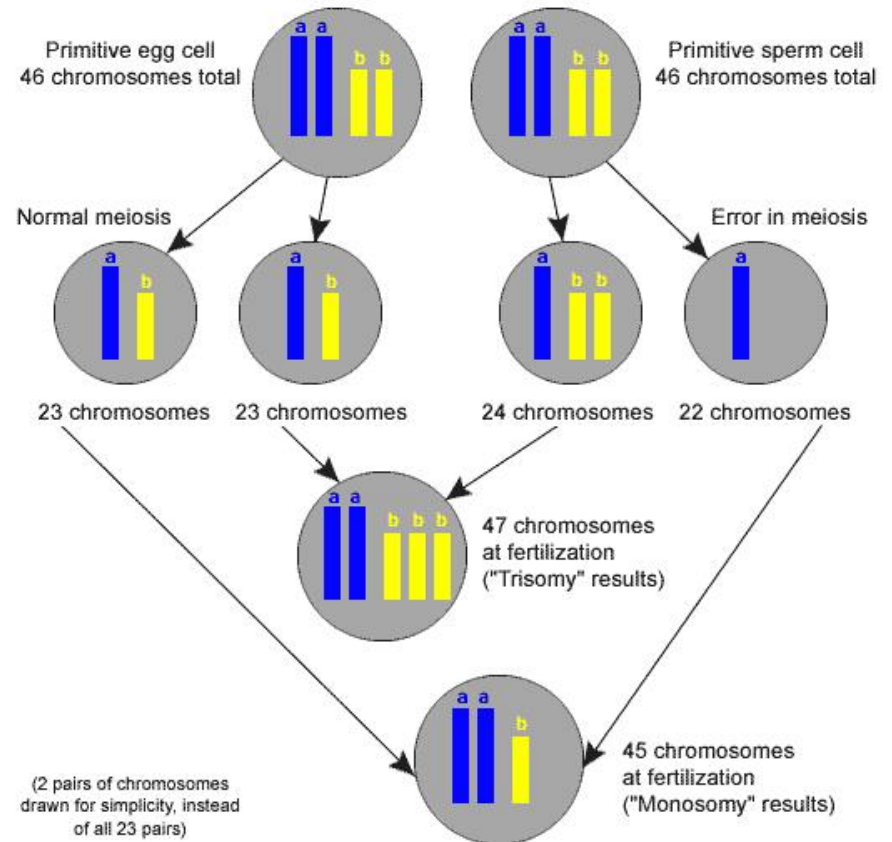
NON-DISJUNCTION

- If it happens in meiosis I, **all** the resulting cells will be affected. If it happens in meiosis II, only **half** will be affected.



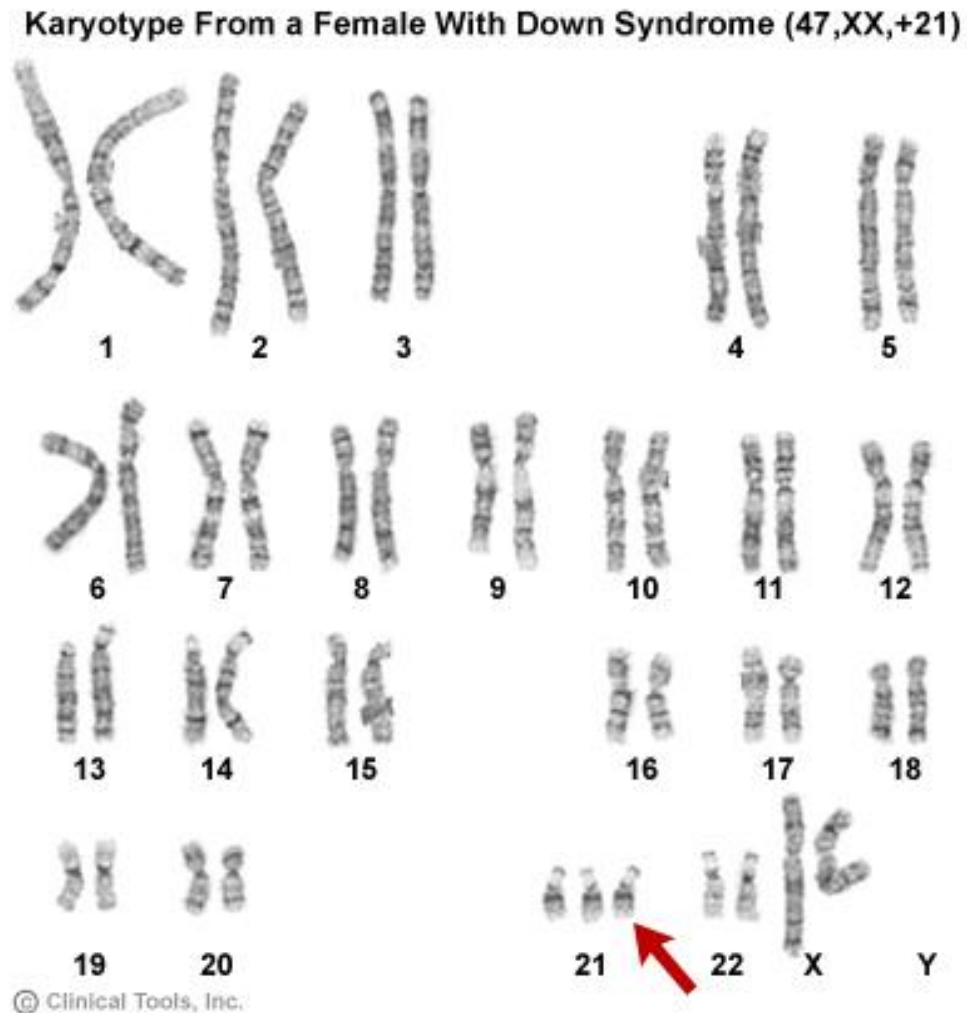
NON-DISJUNCTION

- If a gamete with an extra chromosome is fertilized by a normal gamete, the zygote will have an extra chromosome, called **trisomy**.
- If a gamete missing a chromosome is fertilized by a normal gamete, the zygote will have only one copy of a chromosome, called **monosomy**.



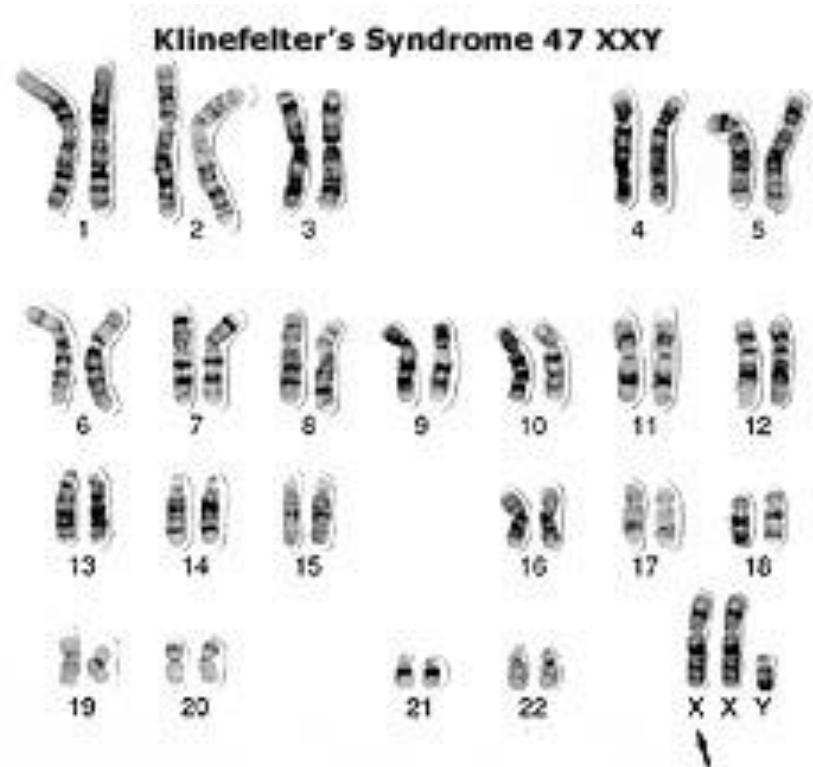
TRISOMY

- Trisomy 21 results in **Down Syndrome**.
- Some effects include mental delay, weakened cardiovascular system, shortened limbs, widely-spaced eyes, receding forehead/chin, and protruding tongue.



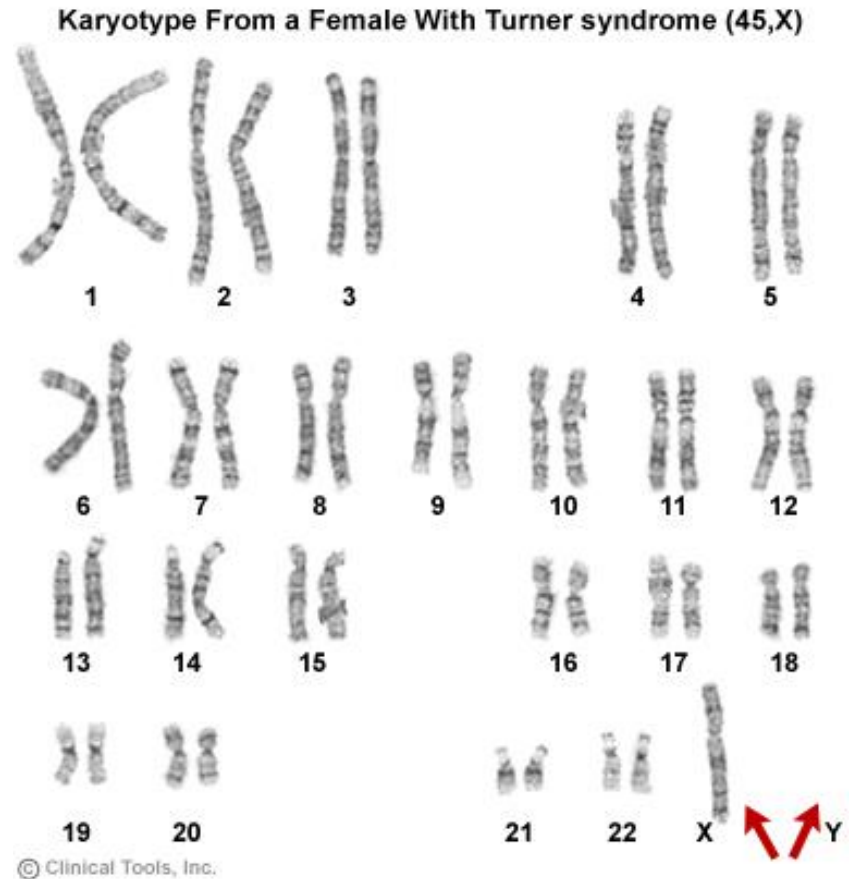
TRISOMY

- **Klinefelter's Syndrome** occurs when an individual receives two X chromosomes and a Y chromosome.
- The result is an infertile male with varying degrees of femininity.



MONOSOMY

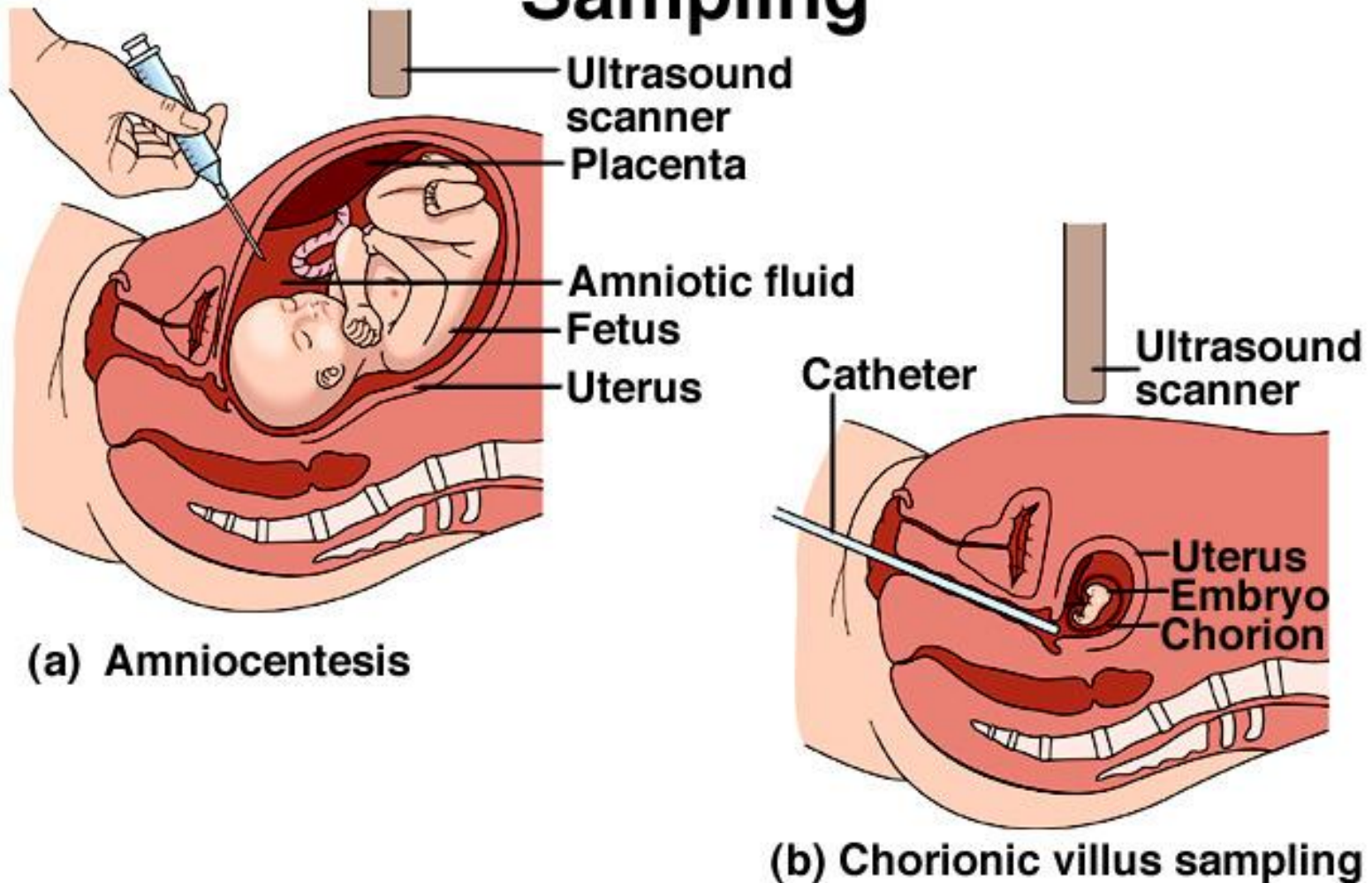
- **Turner Syndrome** occurs when the individual only gets one sex chromosome; an X
- The result is an infertile female with a broad chest, poor breast development, low set ears, short stature and poor hearing amongst other things.



Prenatal testing

Byer/Shainberg/Galliano *Dimensions Of Human Sexuality*, 5e. Copyright © 1999. The McGraw-Hill Companies, Inc. All Rights Reserved.

Amniocentesis & Chorionic Villus Sampling

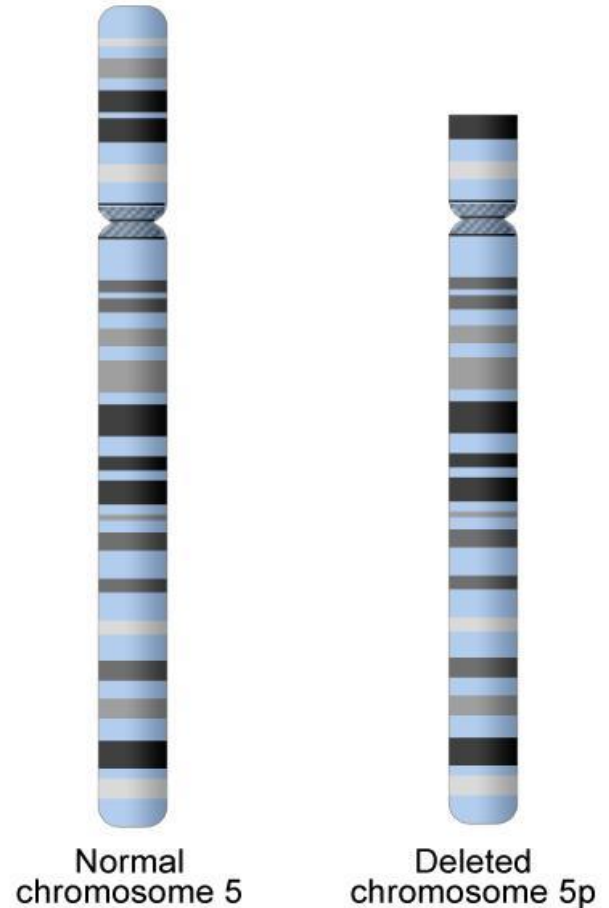


CHANGES IN CHROMOSOMAL STRUCTURE

- **Chromosomal changes can occur spontaneously or from radiation or exposure to certain chemicals.**
- **There are four means of chromosomal changes:**
 - **DELETION**
 - **DUPLICATION**
 - **INVERSION**
 - **TRANSLOCATION**

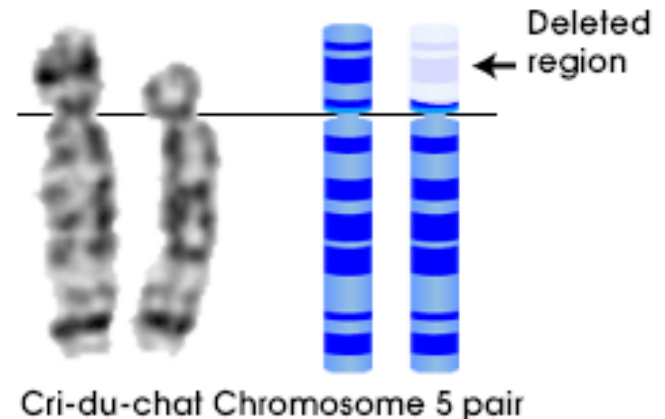
DELETION

- In **deletion**, part of the chromosome is actually lost. Viruses, radiation and chemicals can cause a piece of a chromosome to become dislocated.
- This piece may carry a specific gene which may have a large effect on the host.



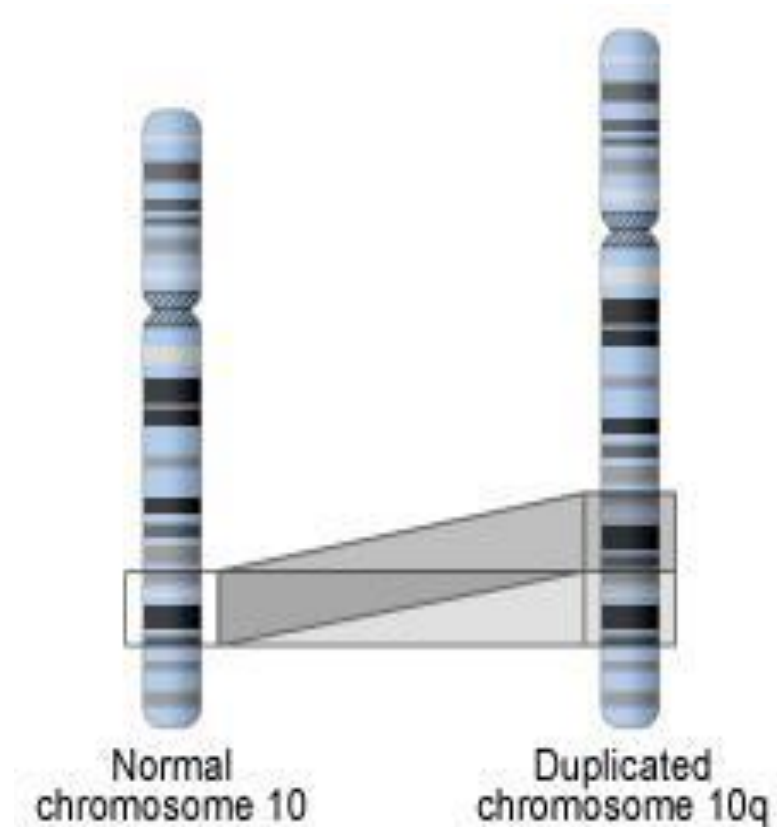
Deletion: Cri du Chat

- “cry of the cat”
- **Deletion of chromosome 5**
- **Symptoms:**
 - small at birth
 - respiratory problems
 - small head (microcephaly)
 - round face, a small chin
 - widely set eyes, folds of skin over their eyes,



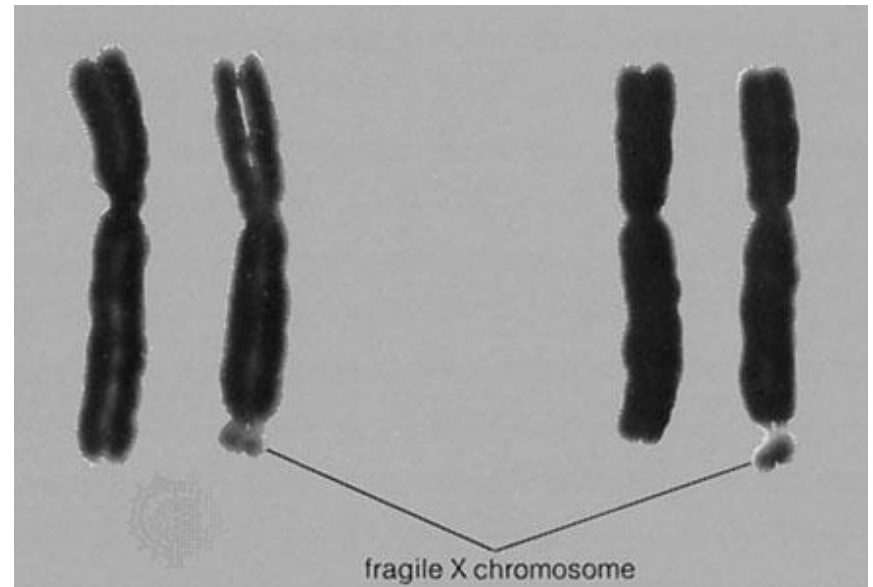
DUPLICATION

- In **duplication**, a gene sequence is repeated one or more times within a chromosome.
- At some point, too many repeats can affect the function of the gene.



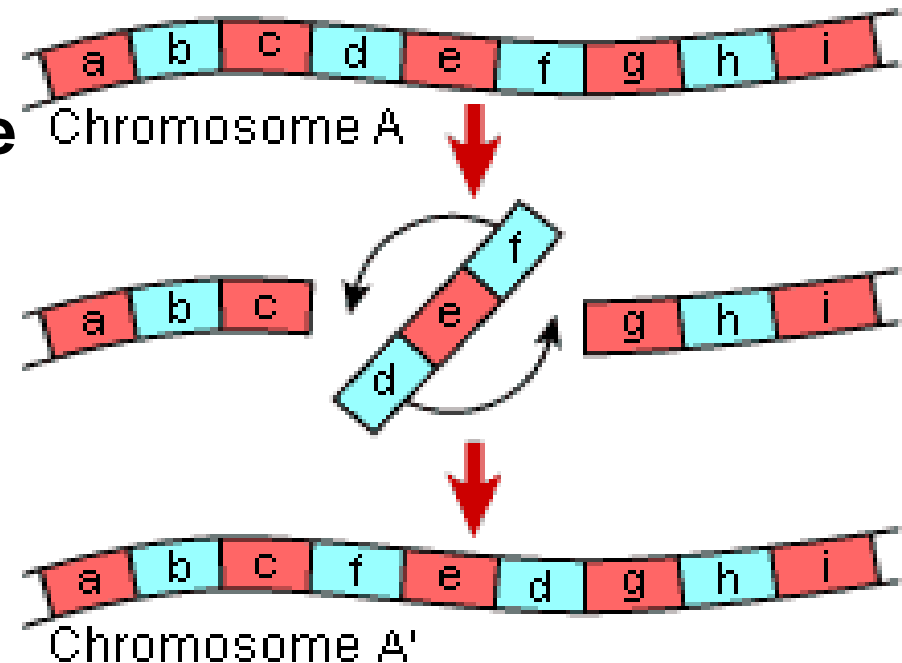
Duplication: Fragile X syndrome

- **Most common form of autism and inherited intellectual disabilities in males**



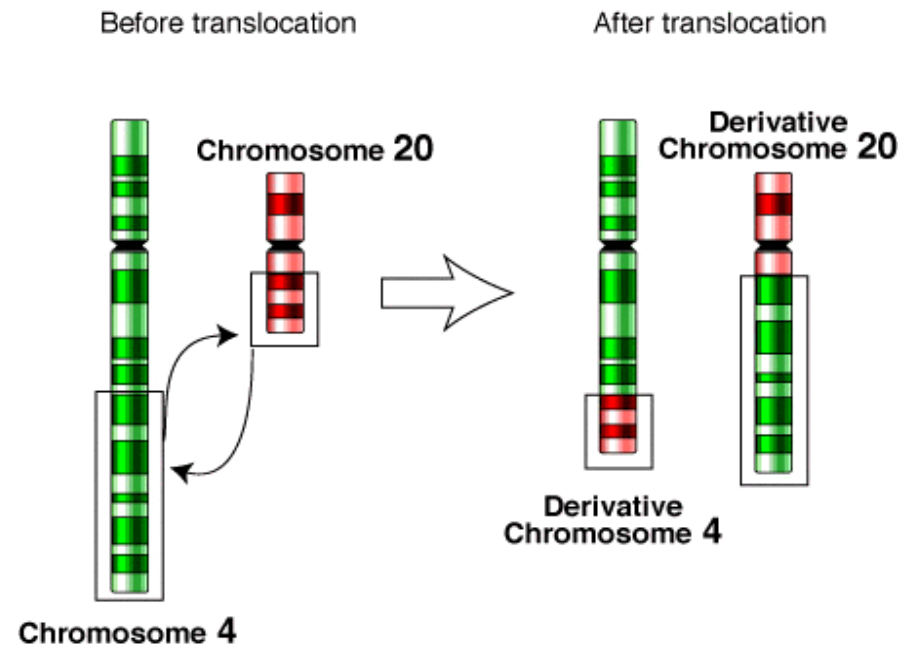
INVERSION

- In **inversion**, a gene segment momentarily becomes free from its chromosome and then reinserts in the **opposite order**.
- This can completely alter the gene's activities.
- **FG syndrome on X chromosome**
 - Effects males
 - Intellectual disabilities



TRANSLOCATION

- In **translocation**, part of a chromosome changes place with a non-homologous chromosome.
- Translocations can result in Chronic Myelogenous Leukemia.

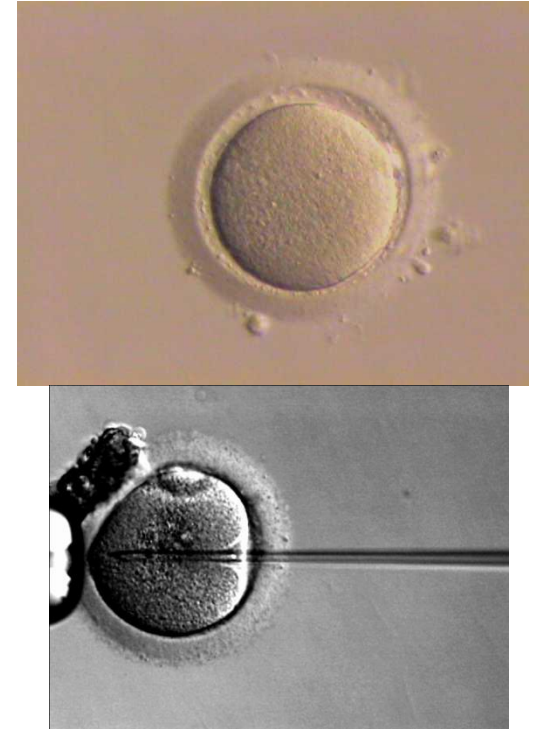


Genetic Technologies

- **In vitro fertilization**
- **Cloning (recombinant DNA technology)**
- **Reproductive cloning (Somatic-cell nuclear transfer)**
- **Stem cells**
- **Transgenic Organisms**

In vitro fertilization

- *In vitro* fertilization in humans is the process of fertilizing egg cells with sperm outside the human body
- The fertilized egg is then transplanted into a human uterus



<http://www.youtube.com/watch?v=D9QDHhnEV9c&feature=related>

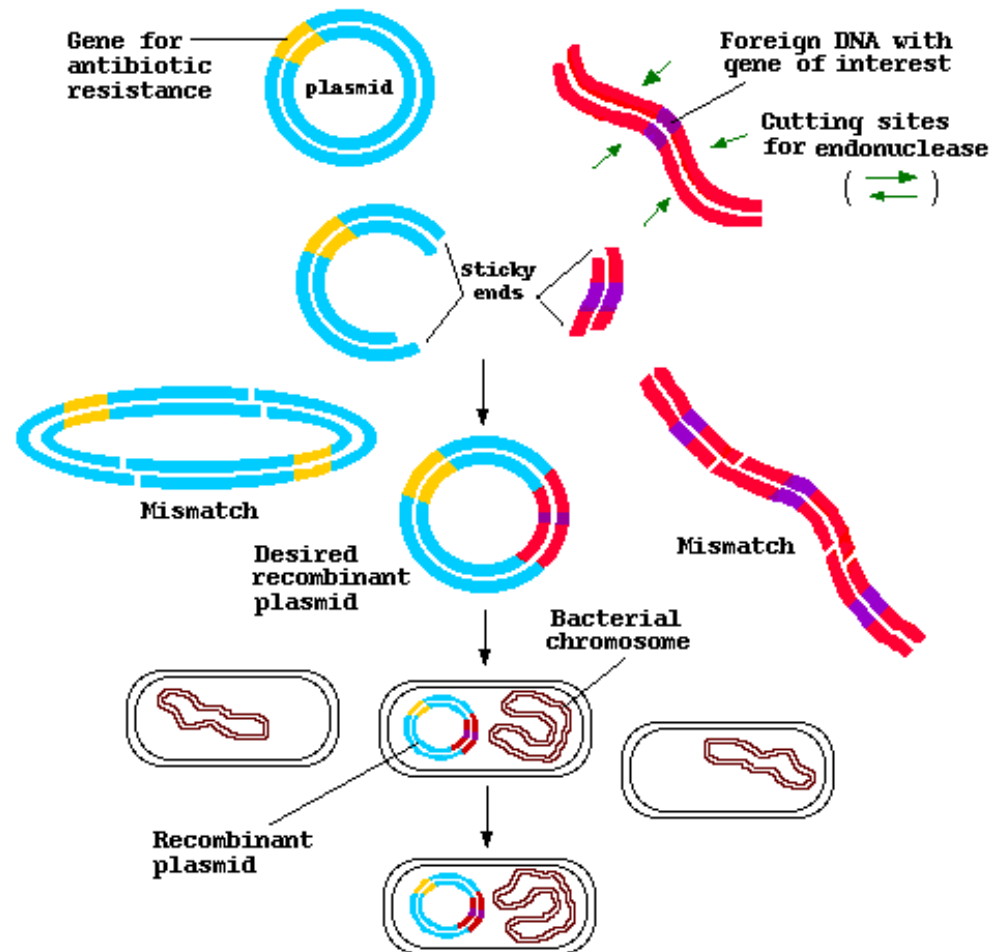
Cloning: Recombinant DNA

- Recombinant DNA techniques allow scientists to equip an organism with DNA that is not normally found within it.
- This DNA causes the cell to make certain proteins.
- But how exactly do scientists make DNA and get it into a cell?

Cloning: Recombinant DNA

- They use a circular molecule of bacterial (*E. coli*) DNA called a plasmid.
- The bacteria (that now contains the DNA) then replicates, creating a DNA “mini-factory” and secreting the desired proteins

Plasmid Insertion



Cloning: Recombinant DNA

- This is actually how they make insulin to treat diabetes!
- This type of genetic modification has also been used in agriculture (and is a source of much debate!)

Reproductive Cloning: Nuclear Cloning

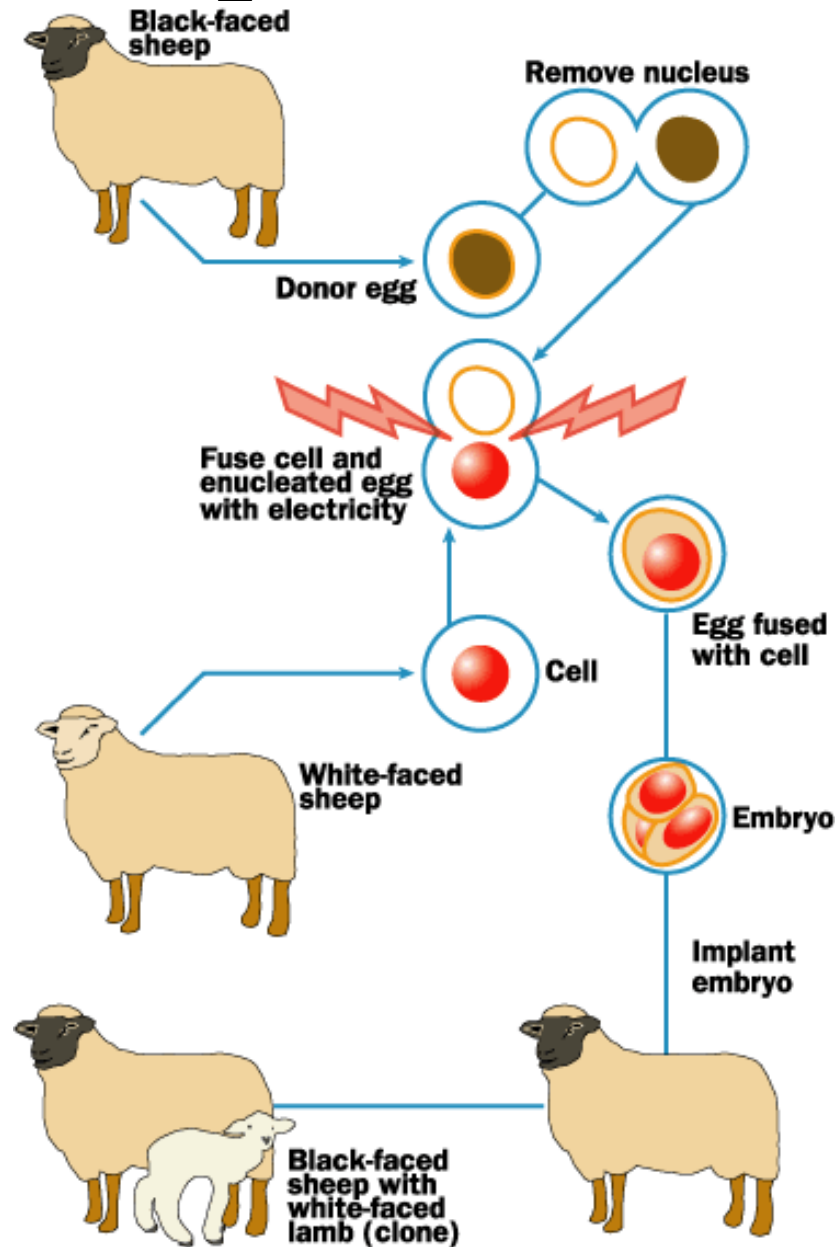
- Producing insulin in *E. coli* is one method of **cloning**.
- What famous sheep comes to mind when you think of cloning?



Cloning: Nuclear Cloning

- Dolly – the first sheep that is genetically identical to its mother!
- Dolly was made by taking an egg cell from a random sheep, removing its nucleus, placing it beside udder cells from Dolly's "mother" and passing an electric charge between the two cells

Cloning: Nuclear Cloning



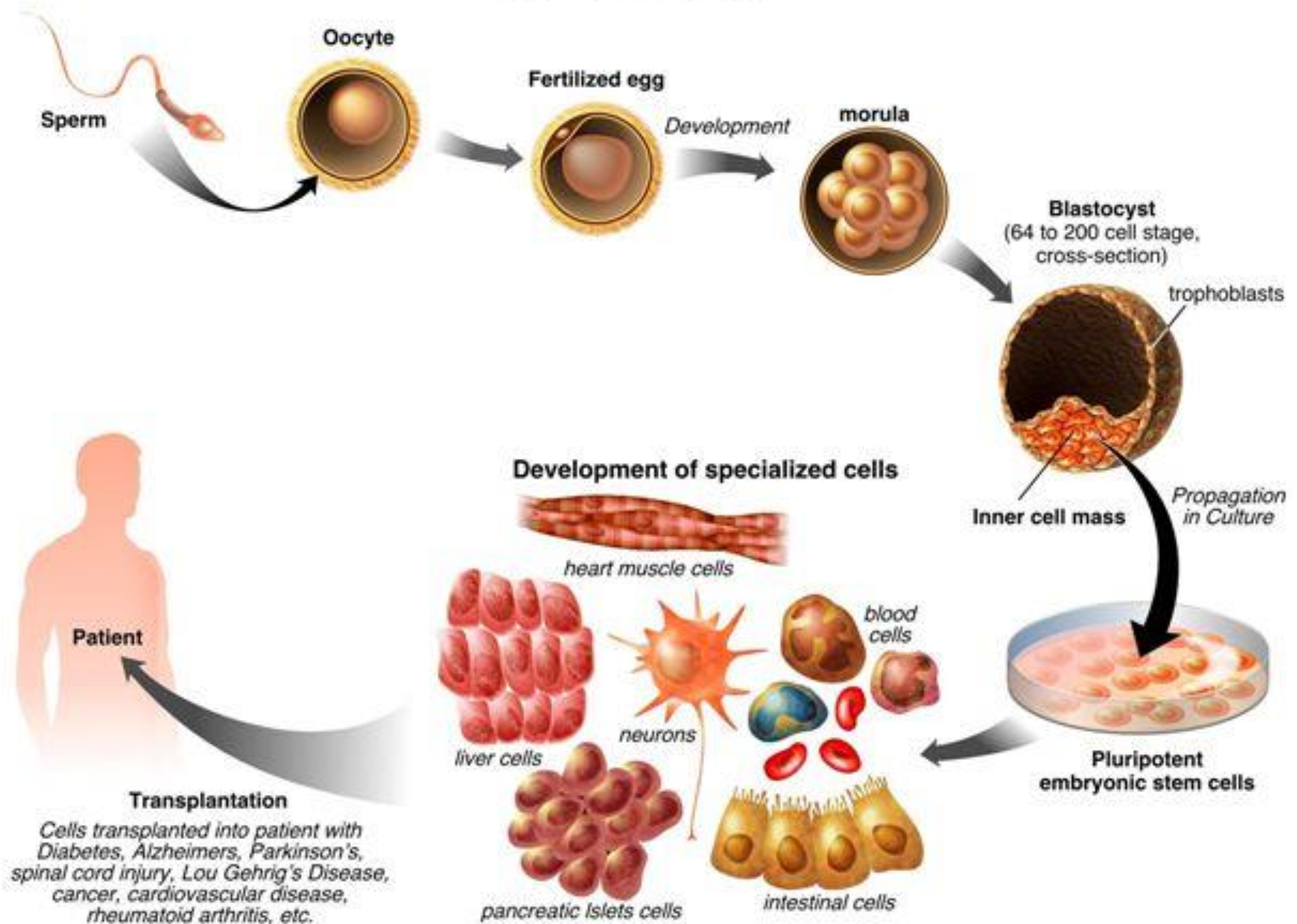
Cloning: Nuclear Cloning

<http://learn.genetics.utah.edu/content/tech/cloning/clickandclone/>

Therapeutic Stem Cells

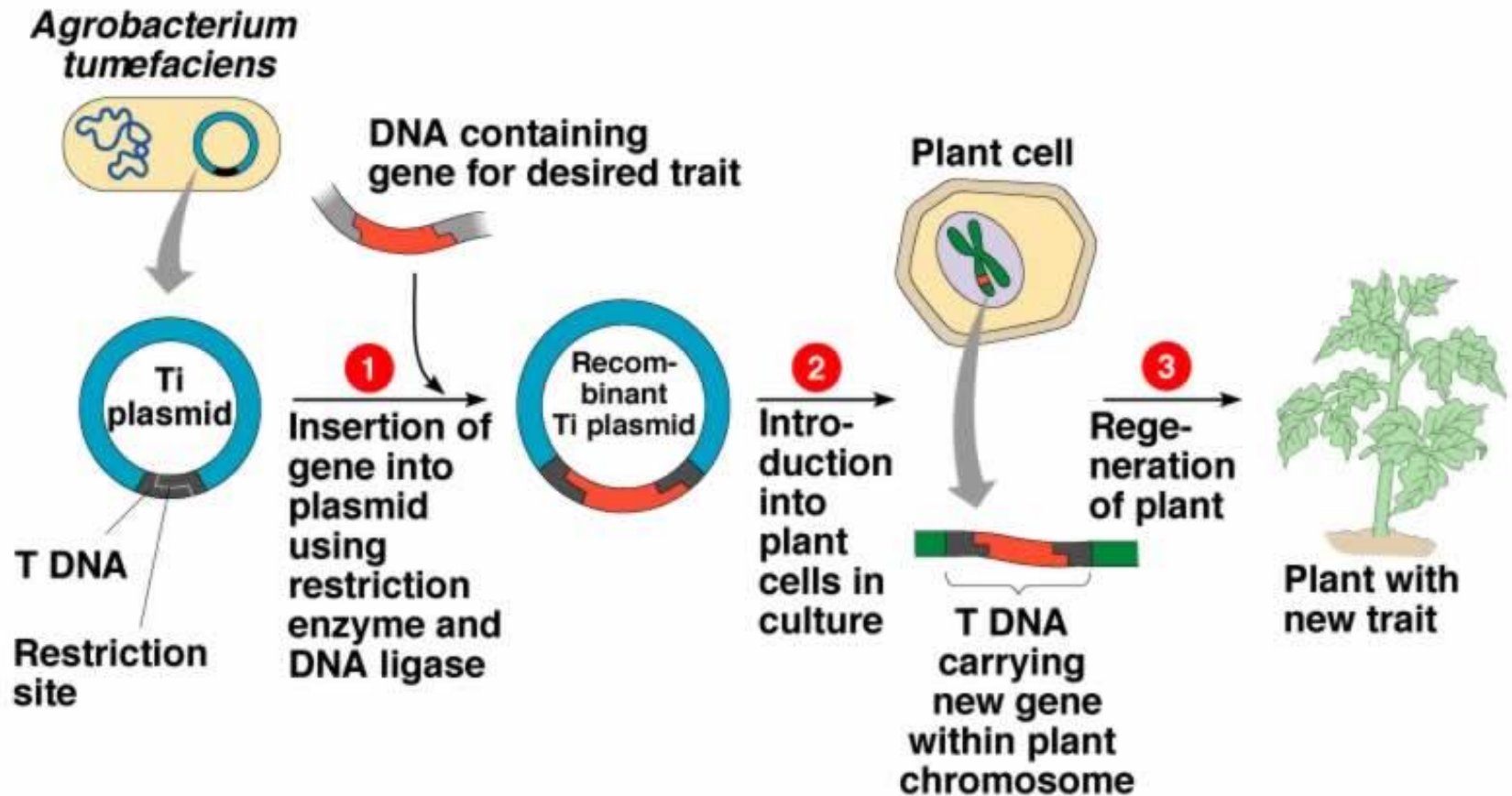
- **A stem cell is an undifferentiated cell that can develop and become specialized into different cell types**
- **Embryonic stem cells**
 - obtained from embryos
- **Adult stem cells**
 - somatic cells that can differentiate into some other cell types
- **Induced pluripotent stem cells**
 - Somatic cells that have been induced to return to a stem-cell-like state

Stem Cell Therapy



Transgenic Plants

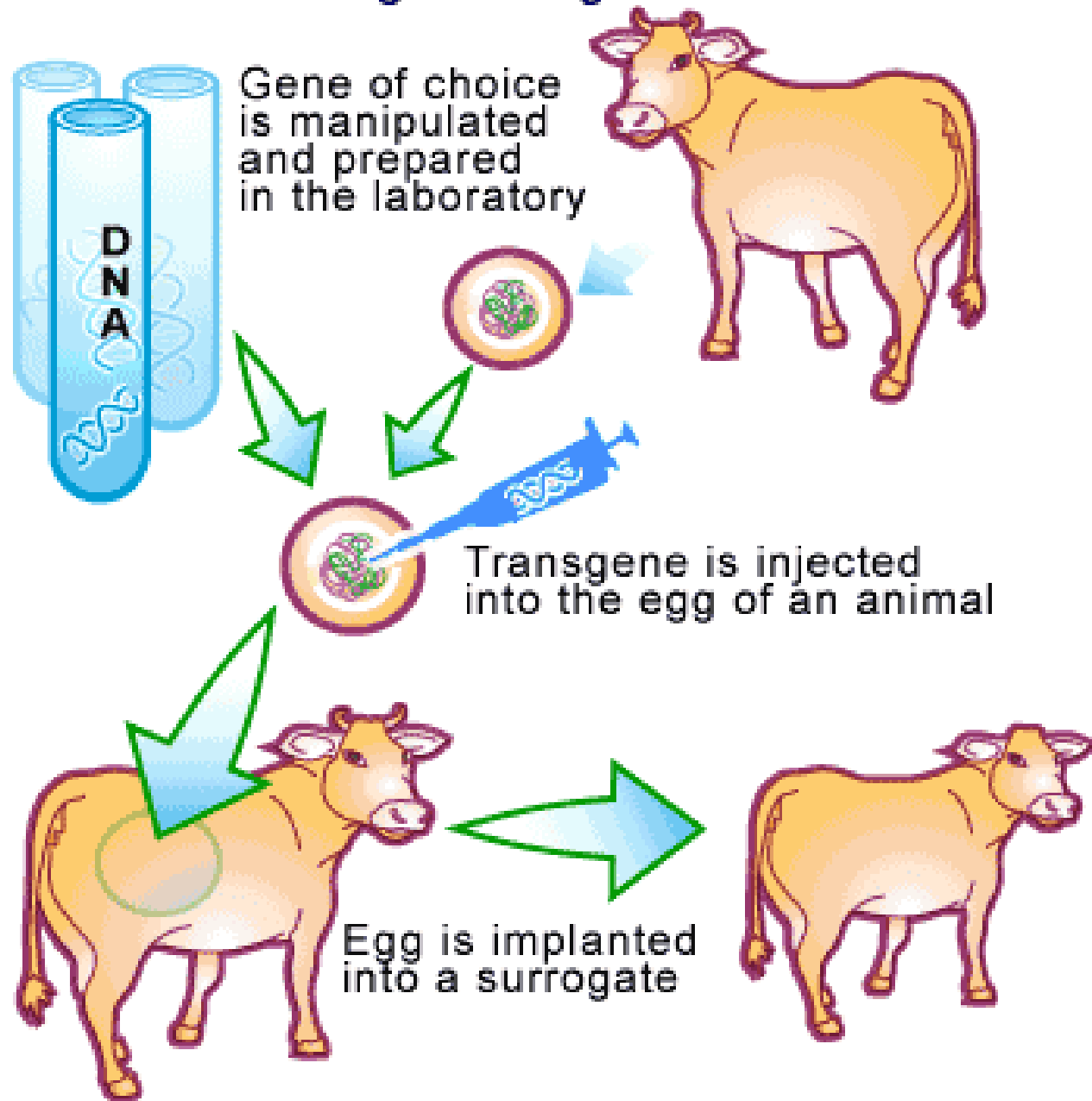
- **Genetically modified (GMO) to express gene products of foreign genes. May include**
 - Herbicide resistance
 - Insect or pest resistance
 - Drought resistance
 - Increased vitamin production
 - Drugs (insulin, vaccines)



Transgenic animals

- **Show slide show**

Creating a transgenic animal



Spider Goats

<http://www.bbc.com/news/science-environment-16554357>

The Ethics of Genetics

- These techniques and practices have the potential to alter the path of evolution in many species (including humans)
- There are also, as you could imagine, highly controversial.

Regulating the use of transgenic organisms

- **Environmental threats**
 - Transfer of GMO genes to other plants
 - Creation of “superweeds”
- **Health effects**
 - Allergy?
- **Social/economic issues**
 - Private enterprise controlling global food markets
 - Ethical concerns